



STAT4 gene

signal transducer and activator of transcription 4

Normal Function

The *STAT4* gene provides instructions for a protein that acts as a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of certain genes. The STAT4 protein is turned on (activated) by immune system proteins called cytokines, which are part of the inflammatory response to fight infection. When activated, the STAT4 protein increases the activity of genes that help immune cells called T-cells mature into specialized T-cells. These specialized T-cells, called Th1 cells, produce specific cytokines and stimulate other immune cells to get rid of foreign invaders (pathogens) in the cell.

Health Conditions Related to Genetic Changes

[juvenile idiopathic arthritis](#)

[rheumatoid arthritis](#)

[systemic lupus erythematosus](#)

[systemic scleroderma](#)

A normal variation in the *STAT4* gene has been associated with an increased risk of developing systemic scleroderma, which is an autoimmune disorder characterized by the buildup of scar tissue (fibrosis) in the skin and internal organs. Although the *STAT4* gene is known to stimulate the immune system in response to pathogens, it is unknown how the gene variation contributes to the increased risk of systemic scleroderma. Researchers believe that a combination of genetic and environmental factors may play a role in development of the condition.

[autoimmune disorders](#)

Studies have associated a normal variation in the *STAT4* gene with an increased risk of several autoimmune disorders. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. These disorders include systemic lupus erythematosus, rheumatoid arthritis, and Sjögren syndrome.

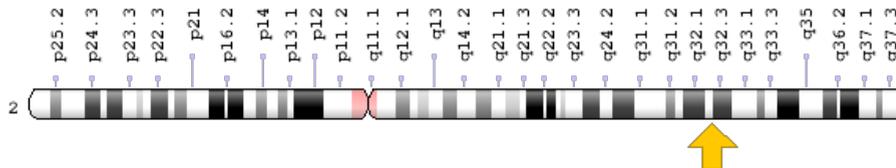
The variant associated with increased risk of autoimmune disorders changes a single DNA building block (nucleotide) in the *STAT4* gene. It is unknown how the gene variation contributes to increased risk of these conditions. Researchers believe that a

combination of genetic and environmental factors may play a role in development of autoimmunity.

Chromosomal Location

Cytogenetic Location: 2q32.2-q32.3, which is the long (q) arm of chromosome 2 between positions 32.2 and 32.3

Molecular Location: base pairs 191,029,576 to 191,172,684 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- SLEB11
- STAT4_HUMAN

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28STAT4%5BTIAB%5D%29+OR+%28signal+transducer+and+activator+of+transcription+4%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

OMIM

- RHEUMATOID ARTHRITIS
<http://omim.org/entry/180300>
- RHEUMATOID ARTHRITIS, SYSTEMIC JUVENILE
<http://omim.org/entry/604302>
- SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION 4
<http://omim.org/entry/600558>

- SJOGREN SYNDROME
<http://omim.org/entry/270150>
- SYSTEMIC LUPUS ERYTHEMATOSUS, SUSCEPTIBILITY TO, 11
<http://omim.org/entry/612253>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_STAT4.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=STAT4%5Bgene%5D>
- HGNC Gene Family: SH2 domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/741>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11365
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6775>
- UniProt
<http://www.uniprot.org/uniprot/Q14765>

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